

Connective Tissue and Vascular Diseases

Disorder	Gene/Exon	Quantity/Material	Frequency Test duration	Method
Ehlers-Danlos Syndrome (EDS) ⁺	<i>COL3A1</i> (CDS complete + MLPA) <i>COL5A1</i> (CDS complete + MLPA) <i>COL5A2</i> (CDS complete)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Familial Thoracic Aortic Aneurysm and Dissection (TAAD) ⁺	<i>ACTA2</i> (CDS complete) <i>COL3A1</i> (CDS complete + MLPA) <i>FBN1</i> (CDS complete + MLPA) <i>MYH11</i> (CDS complete) <i>MYLK</i> (CDS complete) <i>SMAD3</i> (CDS complete) <i>TGFB2</i> (CDS complete) <i>TGFBR1</i> (CDS complete) <i>TGFBR2</i> (CDS complete + MLPA)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA
Marfan Syndrome (MFS) ⁺	<i>FBN1</i> (CDS complete + MLPA) <i>TGFBR1</i> (CDS complete) <i>TGFBR2</i> (CDS complete + MLPA)	5 ml EDTA blood	as needed 4-6 weeks	NGS MLPA

Note:

According to the Genetic Diagnostics Act, every examination request for human genetic diagnostics must be accompanied by a declaration of consent from the patient or his legal representative.

Forms for this as well as request forms for the analyses are available in the download area of our homepage.